

In the Claims:

1. (Previously presented) A method for determining whether a subject is suffering from Schwachman-Diamond Syndrome (SDS) or is an SDS carrier comprising obtaining a nucleic acid sample from the subject, and conducting an assay on the nucleic acid sample to determine the presence or absence of a SBDS gene mutation associated with SDS selected from the group consisting of 183TA>CT, 183TA>CT + 258 + 2T>C, and 258 + 2T>C, and wherein the presence of said SBDS gene mutation associated with SDS in both SBDS alleles indicates that the subject suffers from SDS and the presence of a SBDS gene mutation associated with SDS in one SBDS allele indicates that the subject is an SDS carrier.
2. (Original) The method of claim 1 wherein the assay is selected from the group consisting of probe hybridisation, direct sequencing, restriction enzyme fragment analysis and fragment electrophoretic mobility.
3. (Original) The method of claim 2 wherein the nucleic acid sample is a DNA sample or an RNA sample and the assay is a direct sequencing assay.
4. (Previously presented) The method of claim 3 wherein the nucleic acid sample is a genomic DNA sample and the assay comprises the steps of:
 - (a) amplifying a target portion of the nucleotide sequence of the genomic DNA;
 - (b) obtaining the nucleotide sequence of said amplified target portion; and
 - (c) determining the presence or absence of said SBDS gene mutation associated with SDS in said target portion of the nucleotide sequence.
5. (Previously presented) The method of claim 3 wherein the nucleic acid sample is an RNA sample and the assay comprises the steps of:
 - (a) reverse transcribing the RNA sample to produce a corresponding cDNA;
 - (b) performing at least one polymerase chain reaction with suitable oligonucleotide primers to amplify the SBDS cDNA;
 - (c) obtaining the nucleotide sequence of the amplified SBDS cDNA; and

(d) determining the presence or absence of said SBDS gene mutation associated with SDS in said nucleotide sequence.

6. (Cancelled)

7. (Currently amended) The method of claim 4 or 6 wherein the target portion of the nucleotide sequence is amplified using a primer pair selected from the group consisting of:

- (a) GCGTAAAAAGCCACAATAC (SEQ ID NO:3) and
CTATGACAGTATTCGTAAGACTAGG (SEQ ID NO:4);
- (b) AAATGGTAAGGCAAATACGG (SEQ ID NO:7) and
ACCAAGTTCTTTATTATTAGAAGTGAC (SEQ ID NO:8);
- (c) GCTCAAACCATTACTTACATATTGA (SEQ ID NO:9) and
CACTTGCTTCCATGCAGA (SEQ ID NO:10);
- (d) GCCTTCACTTTCTTCATAGT (SEQ ID NO:31) and
GAAAATATCTGACGTTTACAACA (SEQ ID NO:12);
- (e) GCTTGCCTCAAAGGAAGTT (SEQ ID NO:32) and
CACTCTGGACTTTGCATCTT (SEQ ID NO:14);
- (f) TAAGCCTGCCAGACACAC (SEQ ID NO:19) and
CTATGACAGTATTCGTAAGACTAGG (SEQ ID NO:4);
- (g) AAAGGGTCATTTTAACACTTC (SEQ ID NO:11) and
GAAAATATCTGACGTTTACAACA (SEQ ID NO:12);
- (h) TCCACTGTAGATGTGAACTAACTC (SEQ ID NO:13) and
CACTCTGGACTTTGCATCTT (SEQ ID NO:14); and
- (i) CAGCCGACGACCTTGTTTT (SEQ ID NO:33) and
GTGCCAACGCTGTGTTTT (SEQ ID NO:34).

8. (Original) The method of claim 2 wherein the nucleic acid sample is a DNA sample and the assay is a restriction enzyme fragment analysis.

9. (Original) The method of claim 8 wherein the assay comprises the steps of:

- (a) digesting the DNA with a restriction enzyme to give restriction fragments;

- (b) separating the restriction fragments by agarose gel electrophoresis; and
- (c) detecting the separated fragments by hybridisation of the fragments to a detectably labelled nucleotide probe specific for SBDS.

10. (Previously presented) The method of claim 9, wherein the method is for determining whether a subject is suffering from SDS and wherein the restriction enzyme is at least one of Cac81 and Bsu361.

11. (Previously presented) The method of any one of claims 1 to 10 wherein the subject is a human subject.

12.-20. (Cancelled)

21. (Previously presented) The method of claim 9, wherein the method is for determining whether a subject is an SDS carrier and wherein the restriction enzyme is Nde 1.

22.-53. (Cancelled)